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Dandy-Walker Malformasyonu İle Postaksiyel Polidaktili Birlikteliği: Yenidoğan Olgu Sunumu

Dandy-Walker Malformation with Postaxial Polydactyly: Newborn Case Report

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Öz

Dandy-Walker malformasyonu, serebellar vermis agenezisi veya hipoplazisi, 4. ventrikülün kistik dilatasyonu ve posterior fossa genişlemesi ile karakterize nadir bir konjenital malformasyondur. Hastaların yaklaşık %70-90'ında postnatal hidrosefali gelişir. Dandy-Walker malformasyonun postaksiyel polidaktili ile ilişkisi ise, olası bir otozomal resesif sendrom (OMIM 220220) olarak tanımlanmıştır. Bu vakada Dandy-Walker malformasyonu tanısı alan ve postaksiyel polidaktilisi olan bir infant nadir olması nedeniyle sunuldu.

Anahtar Kelimeler: Dandy-Walker malformasyonu, Polidaktili, Hidrosefali, Apne

Abstract

Dandy-Walker malformation is a rare congenital malformation, characterized by agenesis or hypoplasia of the cerebellar vermis, cystic dilatation of the 4th ventricle and enlargement of the posterior fossa. Postnatal hydrocephalus develops in approximately 70-90% of the patients. The relationship of Dandy-Walker malformation with postaxial polydactyl has been defined as a possible autosomal recessive syndrome (OMIM 220220). Here, we present an infant with postaxial polydactyl accompanying Dandy-Walker malformation because of rare.

Keywords: Dandy-Walker malformation, Polydactyly, Hydrocephalus, Apnea

INTRODUCTION

Dandy-Walker spectrum consists of cystic malformations of the posterior fossa, including the mega cisterna magna, Dandy-Walker malformation (DWM) and Dandy-Walker variant (inferior vermian hypoplasia). Classical DMW is characterized by cystic dilatation of the fourth ventricle, complete or partial agenesis of the cerebellar hemispheres and vermis, and enlargement of the posterior fossa. Its prevalence is approximately 1/30000 live births, accounting for 4-12% of all infant hydrocephalus cases (1). While central nervous system (CNS) anomalies include hydrocephalus, corpus callosum agenesis, polymicrogyria, syringomyelia, holoprosencephaly and occipital encephalocele; congenital heart disease, neural tube defects, cleft palate and cleft lip can be observed as extracranial anomalies (2). The diagnosis of Dandy-Walker malformation can usually be made by fetal or neonatal radiological imaging (3).

In this case report, we presented a neonatal case diagnosed with Dandy-Walker malformation in antenatal ultrasonography (USG) and postnatal cranial magnetic resonance imaging (MRI); developed hydrocephalus in clinical follow-up and accompanied by postaxial polydactyly as a additional anomaly.

CASE REPORT

A 31-year-old father and a 23-year-old mother with epilepsy who were using levetirecetam was born as the first survivor from her first pregnancy and a girl baby with 3085 grams at the 41st gestational week. The APGAR scores were 5 and 7. In physical examination, weight 3085 g (10-25p), height 52 cm (25-50p), head circumference 37 cm (75-90p), body temperature 36.4 ºC,

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respiratory rate 57 / min, pulse 141 / min, blood pressure 64 / 35 mmHg detected. There was hypertelorism in the eyes, frontal bossing, depressed nasal bridge, micrognathia, the 6th finger originating from the metacarpal region in the left hand and a polypoid finger-like appearance in the same area in the right hand (Fig. 1 and 2). Since antenatal USG had an appearance compatible with DWM and a history of hyperechogenic foci in bilateral kidneys, cranial USG revealed a natural width of the ventricular system, cystic dilatation of the 4th ventricle and cerebellar vermis hypoplasia. Cranial MRI showed vermian hypoplasia, enlargement of the 4th ventricle, superior positioning at the level of tentorium cerebelli, and corpus callosum hypoplasia was reported in compatible with DWM (Fig. 3). Parenchymal thinning and nephrocalcinosis were detected in the right kidney on abdominal USG. Submitted urine tests showed calcium / creatinine: 0.02 (normal), cystine: negative, oxalate / creatinine: 126 (0-288), citrate / creatinine: normal. Toxoplasma gondii, rubella, cytomegalovirus, herpes simplex virus infection were negative. The cardiological evaluation was normal. Spontaneous amputation was achieved by tying the accessory fingers of the right and left hand proximally. Chromosome analysis resulted as 46, XY. Hydrocephalus was detected in the brain tomography performed due to an increase of > 1 cm/week in head circumference and a head circumference of > 97 percent in the postnatal 2nd week. Follow-up was recommended to the patient, who was consulted with the Department of Neurosurgery. The patient, who had apneas due to central reasons in the first week follow-up, was discharged on the 15th day of hospitalization, due to the absence of apnea and the ability to be fed orally, to come to the outpatient clinic. Nephrocalcinosis was not detected in the subsequent outpatient clinic follow-ups of the patient, and there was no need for a ventriculoperitoneal shunt.

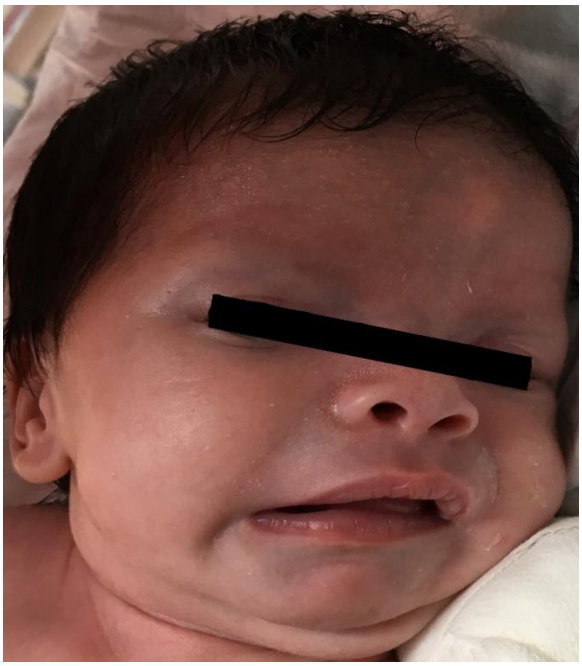

Figure 1. The appearance of facial dysmorphic findings of our case

Figure 2. The view of the 6th finger of our case originating from the metacarpal region in the left hand.

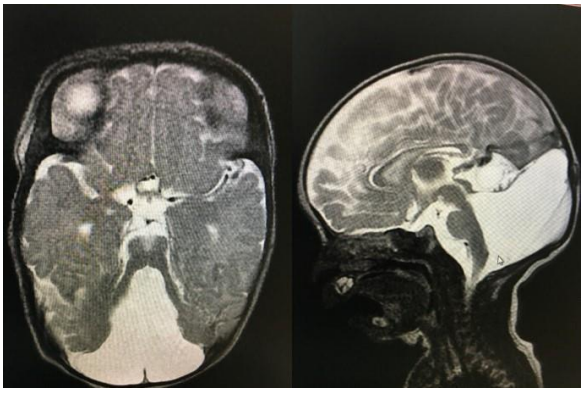

Figure 3. Axial and sagittal MRI: Cystic appearance enlarging the posterior fossa and associated with the 4th ventricle, hypoplastic cerebellum and corpus callosum are observed.

DISCUSSION

Although familial cases have been reported, DWM is thought to be caused by multiple factors. It is known that chromosomal abnormality, other genetic syndromes or teratogens such as alcohol are the causative agents in only a minority of the cases. Although the mechanism of development is not exactly known, it is thought to be related to the process of posterior brain development in weeks intrauterine 7-8th (4). Common clinical signs of Dandy-Walker malformation are characteristic occiput enlargement, macrocrania, mental retardation, cerebellar ataxia, and increased intracranial pressure (5). About a quarter of these patients show signs during the neonatal period. Concomitant congenital anomalies are craniofacial, cardiac, renal and skeletal anomalies. Cerebral anomalies can cause severe respiratory failure such as apneic breathing (6). Our case also had craniofacial dysmorphic findings and additionally postaxial polydactyly. Although he did not have hydrocephalus in the first postnatal days, he developed hydrocephalus in his follow-up, but a ventriculoperitoneal shunt was not needed. Nephrocalcinosis was detected as a renal finding, and stone examinations sent for differential diagnosis were found to be normal. Recurrent apnea attacks were thought to be due to respiratory center involvement.

The combination of Dandy-Walker malformation and postal polydactyly (OMIM 220220, 2012) was first described by Pierquin et al. (1989) in fetuses without other malformations. Cavalcanti and Salomao (1999) also described a male baby born to consanguineous parents with similar characteristics and suggested that there was autosomal recessive inheritance. In our case, on the other hand, there was no consanguinity between the parents and no similar disease was observed in the family before.

Dandy-Walker malformation and polydactyly may be isolated or associated with other malformations. However, differential diagnosis is usually limited. In Mohr syndrome (mouth-face-finger syndrome type II), central nervous system features may include DWM (OMIM 252100, 2012), hydrocephalus, and/or porencephaly. This disorder typically includes preaxial polydactyly of the hands and postaxial polydactyly of the feet; also, lobulated tongue and alveolar cleft are easily distinguishing features (7).

Patients with Ritscher-Schinzel syndrome or 3C (Cranio-cerebello-cardiac) syndrome (OMIM: 220210) have Dandy-Walker-like malformation and atrioventricular septal defect (8). Papadopoulou et al. (2005) described a male patient with 3C syndrome with posterior embryotoxone. However, postaxial polydactyly was not described in any of the patients reported with 3C syndrome.

Recently renamed Joubert syndrome and related disorders, Joubert syndrome (OMIM: 213300) was redefined to include all syndromes showing 'molar tooth sign' (including Meckel syndrome). In this syndrome, there is long midbrain, dysplastic caudal medulla and vermis hypoplasia or dysplasia (9). These features include tachypnea episodes, apnea episodes, and irregular jerky eye movements that occur during waking and non-REM sleep. These findings were not observed in our case. Also, MRI brain images did not show the molar tooth sign required for diagnosis (10).

Conclusion

Due to its multi-systemic involvement, the association of Dandy-Walker malformation and postaxial polydactyly is important for the diagnosis and early detection of conditions such as hydrocephalus that may develop during follow-up. This diagnosis should be considered especially in patients with agenesis or hypoplasia of cerebellar vermis, cystic dilatation of the 4th ventricle, enlargement of the posterior fossa and polydactyly, and a multidisciplinary approach is required in its treatment.

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